

CLAIMS

What is claimed is:

- 5 1. A method for detecting aberrant promoter methylation associated with
predisposition for cancers of the breast, lung, and colon, in a human comprising detecting
methylation of the PAX5 α gene.
2. The method of claim 1 further comprising the steps of:
10 expanding the number of copies of the PAX5 α gene by using a polymerase
chain reaction to amplify a portion of the gene where the promoter methylation resides,
thereby generating an amplification product; and
 using an aliquot of the amplification product generated by the first
polymerase chain reaction in a second, methylation-specific polymerase chain reaction to
15 detect the presence of inactivation of the PAX5 α gene by methylation.
3. A method for detecting aberrant promoter methylation associated with
predisposition for cancers of the breast, lung, and colon, in a human comprising detecting
methylation of the PAX5 β gene.

4. The method of claim 3 further comprising the steps of:
- expanding the number of copies of the PAX5 β gene by using a polymerase chain reaction to amplify a portion of the gene where the promoter methylation resides,
- 5 thereby generating an amplification product; and
- using an aliquot of the amplification product generated by the first polymerase chain reaction in a second, methylation-specific polymerase chain reaction to detect the presence of inactivation of the PAX5 β gene by methylation.
- 10 5. A method of monitoring for cancer in a human, comprising detecting gene inactivation in a biological fluid by ascertaining the presence of gene-specific promoter methylation in the cells of the biological fluid, and further comprising the steps of:
- obtaining a sample the biological fluid containing the PAX5 α gene,
- wherein the step of obtaining a sample comprises the step of selecting a member from the
- 15 group consisting of plasma, mucus, fecal stool, and sputum;
- expanding the number of copies of the PAX5 α gene by using a polymerase chain reaction to amplify a portion of the gene where the promoter methylation resides,
- thereby generating an amplification product; and
- using an aliquot of the amplification product generated by the first
- 20 polymerase chain reaction in a second, methylation-specific, polymerase chain reaction to detect the presence of inactivation of the PAX5 α gene in the biological fluid.

6. A method of monitoring for cancer in a human, comprising detecting gene inactivation in a biological fluid by ascertaining the presence of gene-specific promoter methylation in the cells of the biological fluid, and further comprising the steps of:

obtaining a sample the biological fluid containing the PAX5 β gene,

5 wherein the step of obtaining a sample comprises the step of selecting a member from the group consisting of plasma, mucus, fecal stool, and sputum;

expanding the number of copies of the PAX5 β gene by using a polymerase chain reaction to amplify a portion of the gene where the promoter methylation resides, thereby generating an amplification product; and

10 using an aliquot of the amplification product generated by the first polymerase chain reaction in a second, methylation-specific, polymerase chain reaction to detect the presence of inactivation of the PAX5 β gene in the biological fluid.

7. A method of monitoring for cancer in a human, comprising detecting gene
15 inactivation in a biological fluid by ascertaining the presence of gene-specific promoter methylation in the cells of the biological fluid, and further comprising the steps of:

subjecting DNA in the biological fluid to bisulfite modification;

expanding the number of copies of PAX5 α gene in the DNA by using
primer sequences which recognize the bisulfite-modified DNA template, but which not
20 discriminate between methylated and unmethylated alleles, in a polymerase chain reaction to amplify a CpG-rich portion of the PAX5 α gene where the promoter methylation resides, thereby generating an amplification product containing fragments of the PAX5 α gene;

using an aliquot of the amplification product generated by the first polymerase chain reaction in a second, methylation-specific, polymerase chain reaction employing primer sequences specific to a methylated DNA template to detect the presence of inactivation of the PAX5 α gene.

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8. A single-stranded DNA primer for determination of a nucleotide sequence of a PAX5 α gene or for use in a polymerase chain reaction wherein said primer comprises a sequence selected from the group consisting of: (i) SEQ ID NO:1 or a complement thereof, (ii) SEQ ID NO:2 or a complement thereof, (iii) SEQ ID NO:5 or a complement thereof, and (iv) SEQ ID NO:6 or a complement thereof.

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9. A single-stranded DNA primer for determination of a nucleotide sequence of a PAX5 β gene or for use in a polymerase chain reaction wherein said primer comprises a sequence selected from the group consisting of: (i) SEQ ID NO:3 or a complement thereof, (ii) SEQ ID NO:4 or a complement thereof, (iii) SEQ ID NO:7 or a complement thereof, and (iv) SEQ ID NO:8 or a complement thereof.

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